cystic fibrosis

Cystic fibrosis is an inherited disease characterized by the buildup of thick, sticky mucus that can damage many of the body's organs. The disorder's most common signs and symptoms include progressive damage to the respiratory system and chronic digestive system problems. The features of the disorder and their severity varies among affected individuals.

Mucus is a slippery substance that lubricates and protects the linings of the airways, digestive system, reproductive system, and other organs and tissues. In people with cystic fibrosis, the body produces mucus that is abnormally thick and sticky. This abnormal mucus can clog the airways, leading to severe problems with breathing and bacterial infections in the lungs. These infections cause chronic coughing, wheezing, and inflammation. Over time, mucus buildup and infections result in permanent lung damage, including the formation of scar tissue (fibrosis) and cysts in the lungs.

Most people with cystic fibrosis also have digestive problems. Some affected babies have meconium ileus, a blockage of the intestine that occurs shortly after birth. Other digestive problems result from a buildup of thick, sticky mucus in the pancreas. The pancreas is an organ that produces insulin (a hormone that helps control blood sugar levels). It also makes enzymes that help digest food. In people with cystic fibrosis, mucus blocks the ducts of the pancreas, reducing the production of insulin and preventing digestive enzymes from reaching the intestines to aid digestion. Problems with digestion can lead to diarrhea, malnutrition, poor growth, and weight loss. In adolescence or adulthood, a shortage of insulin can cause a form of diabetes known as cystic fibrosis-related diabetes mellitus (CFRDM).

Cystic fibrosis used to be considered a fatal disease of childhood. With improved treatments and better ways to manage the disease, many people with cystic fibrosis now live well into adulthood. Adults with cystic fibrosis experience health problems affecting the respiratory, digestive, and reproductive systems. Most men with cystic fibrosis have congenital bilateral absence of the vas deferens (CBAVD), a condition in which the tubes that carry sperm (the vas deferens) are blocked by mucus and do not develop properly. Men with CBAVD are unable to father children (infertile) unless they undergo fertility treatment. Women with cystic fibrosis may experience complications in pregnancy.

Frequency

Cystic fibrosis is a common genetic disease within the white population in the United States. The disease occurs in 1 in 2,500 to 3,500 white newborns. Cystic fibrosis is less

common in other ethnic groups, affecting about 1 in 17,000 African Americans and 1 in 31,000 Asian Americans.

Genetic Changes

Mutations in the *CFTR* gene cause cystic fibrosis. The *CFTR* gene provides instructions for making a channel that transports negatively charged particles called chloride ions into and out of cells. Chloride is a component of sodium chloride, a common salt found in sweat. Chloride also has important functions in cells; for example, the flow of chloride ions helps control the movement of water in tissues, which is necessary for the production of thin, freely flowing mucus.

Mutations in the *CFTR* gene disrupt the function of the chloride channels, preventing them from regulating the flow of chloride ions and water across cell membranes. As a result, cells that line the passageways of the lungs, pancreas, and other organs produce mucus that is unusually thick and sticky. This mucus clogs the airways and various ducts, causing the characteristic signs and symptoms of cystic fibrosis.

Other genetic and environmental factors likely influence the severity of the condition. For example, mutations in genes other than *CFTR* might help explain why some people with cystic fibrosis are more severely affected than others. Most of these genetic changes have not been identified, however.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- CF
- cystic fibrosis of pancreas
- fibrocystic disease of pancreas
- mucoviscidosis

Diagnosis & Management

Formal Diagnostic Criteria

- ACT Sheet: Elevated IRT +/- DNA
  https://www.ncbi.nlm.nih.gov/books/NBK55827/bin/CF.pdf
- American College of Obstetricians and Gynecologists Committee on Genetics.. ACOG Committee Opinion No. 486: Update on carrier screening for cystic fibrosis. Obstet Gynecol. 2011 Apr;117(4):1028-31. doi: 10.1097/AOG.0b013e31821922c2. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/21422883
- Grody WW, Thompson BH, Gregg AR, Bean LH, Monaghan KG, Schneider A, Lebo RV. ACMG position statement on prenatal/preconception expanded carrier screening. Genet Med. 2013 Jun;15(6):482-3. doi: 10.1038/gim.2013.47.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/23619275
- National Guideline Clearinghouse: Molecular testing for cystic fibrosis carrier status practice guidelines: recommendations of the National Society of Genetic Counselors.
  https://www.guideline.gov/summaries/summary/47400/molecular-testing-for-cystic-fibrosis-carrier-status-practice-guidelines-recommendations-of-the-national-society-of-genetic-counselors?q=cystic+fibrosis

Formal Treatment/Management Guidelines

- Cochrane Reviews: Cystic Fibrosis
  http://www.cochranelibrary.com/topic/Genetic%20disorders/Cystic%20fibrosis/
- National Guideline Clearinghouse: Clinical practice guidelines from the Cystic Fibrosis Foundation for preschoolers with cystic fibrosis.
  https://www.guideline.gov/summaries/summary/50221/clinical-practice-guidelines-from-the-cystic-fibrosis-foundation-for-preschoolers-with-cystic-fibrosis
- National Guideline Clearinghouse: Cystic fibrosis pulmonary guidelines. Chronic medications for maintenance of lung health. (Cystic Fibrosis Foundation)
  https://www.guideline.gov/summaries/summary/45307/cystic-fibrosis-pulmonary-guidelines-chronic-medications-for-maintenance-of-lung-health

Genetic Testing

- Genetic Testing Registry: Cystic fibrosis
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0010674/

Other Diagnosis and Management Resources

- American Society for Reproductive Medicine: Male Infertility
  http://www.asrm.org/topics/detail.aspx?id=1331
- Baby's First Test
  http://www.babysfirsttest.org/newborn-screening/conditions/cystic-fibrosis-cf
- GeneReview: Cystic Fibrosis and Congenital Absence of the Vas Deferens
  https://www.ncbi.nlm.nih.gov/books/NBK1250
- Genomics Education Programme (UK)
  https://www.genomicseducation.hee.nhs.uk/resources/genetic-conditions-factsheets/item/74-cystic-fibrosis
- MedlinePlus Encyclopedia: Cystic Fibrosis
  https://medlineplus.gov/ency/article/000107.htm

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Cystic Fibrosis
  https://medlineplus.gov/ency/article/000107.htm
- Health Topic: Cystic Fibrosis
  https://medlineplus.gov/cysticfibrosis.html
- Health Topic: Newborn Screening
  https://medlineplus.gov/newbornscreening.html

Genetic and Rare Diseases Information Center

- Cystic fibrosis
  https://rarediseases.info.nih.gov/diseases/6233/cystic-fibrosis

Additional NIH Resources

- GeneEd
  https://geneed.nlm.nih.gov/topic_subtopic.php?tid=142&sid=146
- National Heart, Lung, and Blood Institute
  https://www.nhlbi.nih.gov/health/health-topics/topics/cf/
- National Human Genome Research Institute
  https://www.genome.gov/10001213/

Educational Resources

- Boston Children's Hospital
  http://www.childrenshospital.org/conditions-and-treatments/conditions/cystic-fibrosis
- Centre for Genetics Education
  http://www.genetics.edu.au/Publications-and-Resources/Genetics-Fact-Sheets/FS41CYSTICFIBROSIS.pdf
- CFTR2: Clinical and Functional Translation of CFTR
  https://cftr2.org/
- Disease InfoSearch: Cystic Fibrosis
  http://www.diseaseinfosearch.org/Cystic+Fibrosis/2071
- Emory University School of Medicine: Cystic Fibrosis Carrier Testing
  http://genetics.emory.edu/documents/resources/Emory_Human_Genetics_Cystic_Fibrosis_Carrier.PDF
- Genetic Science Learning Center, University of Utah
  http://learn.genetics.utah.edu/content/disorders/singlegene/
- KidsHealth from the Nemours Foundation
  http://kidshealth.org/en/kids/cystic-fibrosis.html
- MalaCards: cystic fibrosis
  http://www.malacards.org/card/cystic_fibrosis
- March of Dimes
  http://www.marchofdimes.org/baby/cystic-fibrosis-and-your-baby.aspx
- Merck Manual Consumer Version
  http://www.merckmanuals.com/home/children-s-health-issues/cystic-fibrosis-cf/cystic-fibrosis
- My46 Trait Profile
  https://www.my46.org/trait-document?trait=Cystic%20fibrosis&type=profile
- Orphanet: Cystic fibrosis
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=586

- The Cleveland Clinic Health Information Center
  http://my.clevelandclinic.org/health/articles/cystic-fibrosis
- Virginia Department of Health
  http://www.vdh.virginia.gov/content/uploads/sites/33/2016/11/Parent-Fact-Sheet_CF_English.pdf
- Your Genes Your Health from Cold Spring Harbor Laboratory
  http://www.ygyh.org/cf/whatisit.htm

Patient Support and Advocacy Resources

- American Lung Association
  http://www.lung.org/lung-health-and-diseases/lung-disease-lookup/cystic-fibrosis/
- Canadian Cystic Fibrosis Foundation
  http://www.cysticfibrosis.ca/
- Cystic Fibrosis Foundation
  https://www.cff.org/
- Cystic Fibrosis Research, Inc.
  http://www.cfri.org/
- Cystic Fibrosis Trust (UK)
  https://www.cysticfibrosis.org.uk/
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/cystic-fibrosis/
- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/cystic_f.html

GeneReviews

- Cystic Fibrosis and Congenital Absence of the Vas Deferens
  https://www.ncbi.nlm.nih.gov/books/NBK1250

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22cystic+fibrosis%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Cystic+Fibrosis%5BMAJR%5D%29+AND+%28cystic+fibrosis%5BTI%5D%29+AND+review%5Bpt%5D+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D

OMIM

- CYSTIC FIBROSIS
  http://omim.org/entry/219700

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  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2662914/
- Gardner J. What you need to know about cystic fibrosis. Nursing. 2007 Jul;37(7):52-5. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17603376
- GeneReview: Cystic Fibrosis and Congenital Absence of the Vas Deferens
  https://www.ncbi.nlm.nih.gov/books/NBK1250
- Gershman AJ, Mehta AC, Infeld M, Budev MM. Cystic fibrosis in adults: an overview for the internist. Cleve Clin J Med. 2006 Dec;73(12):1065-74. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17190310
- Merlo CA, Boyle MP. Modifier genes in cystic fibrosis lung disease. J Lab Clin Med. 2003 Apr; 141(4):237-41. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12677168
- Ratjen F, Döring G. Cystic fibrosis. Lancet. 2003 Feb 22;361(9358):681-9. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12606185
- Rowe SM, Miller S, Sorscher EJ. Cystic fibrosis. N Engl J Med. 2005 May 12;352(19):1992-2001.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15888700

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